FONDATION Maladies Rares

LES LAURÉATS DE L'APPEL À PROJETS 'GENOMICS'

MARS 2020

RESPONSABLE DU PROJET	VILLE	APPROCHE TECHNOLOGIQUE	TITRE DU PROJET
Benoit Arveiler	Bordeaux	Exomes	Search for new albinism genes.
Julien Barc	Nantes	RNAseq	Molecular mechanism of a new syndrome characterized by cardiac electrical and developmental defects and associated with an enhancer deletion of chromosome 4q25
Marion Delous	Lyon	Génomes	Towards the identification of the genetic causes of rare autosomal dominant forms of idiopathic scoliosis
Alexandre Fabre	Marseille	Génomes	Identification of new genes associated with syndromic congénital diarrhea syndrom
Laura Mary	Rennes	Exomes	Identification of genes involved in syndromic disorders of sex development
François-Xavier Mauvais	Paris	RNAseq	Identifying the molecular basis underlying the heterogeneity among the spectrum of lysosomal acid lipase deficiency by a proteogenomic approach supported by bioinformatics
Sylvie Mazoyer	Lyon	RNAseq	A transcriptomic study in zebrafish models of RNU4A-TAC-associated rare diseases: connecting U12 splicing defects to developmental abnormalities
Pierre Ronco	Paris	Génomes	Whole Genome Sequencing to Unravel the Genetic Mechanisms of PLA2R-Associated Membranous Nephropathy
Caroline Schluth-Bolard	Lyon	HiC	Characterization of 3D chromatin architecture disruption in chromosomal rearrangements to identify candidate genes in neurodevelopmental disorders
Julie Steffann	Paris	RNAseq	Does nuclear transfer alter mitochondrial-nuclear crosstalks in the human preimplantation embryo?
Pierre-Louis Tharaux	Paris	ScRNAseq	TRAJHISTORY projet: assessing single cell TRAJectories and alterations of cell communication and tissue HISTOry to get causal insights into rare and catastrophic glomerular diseases
Ha Trang	Paris	Génomes	Congenital Central Hypoventilation Syndrome - searching causal genes in patients without PHOX2B mutations